

4150 Cousins: What 7 DNA Ancestry Tests Can Tell You About Your Kin

by

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Author's Declaration

I hereby declare that I am the sole author of this research paper.

I authorize the University of Waterloo to lend this research paper to other institutions or individuals for the purpose of scholarly research.

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Abstract

This thesis presents the results of seven commercialized DNA ancestry tests that are all available to the public, for under \$400 Canadian dollars each. This research is conducted to explore the use of commercialized DNA ancestry tests. The results from each test are compared in order to determine what they are able to tell a customer. The tests used are not the only tests available, but are chosen because of their popularity, price, and what they claim to be able to report to their customer. I find the databases that the tests include online to ‘find relatives’, who are other customers having the same Haplogroup or another matching genetic identifier, to be the most troublesome aspect of the results. Specifically, it is important for the public to clearly understand that these tests are not as conclusive as they are advertised to be, so that they are not misled in thinking that the tests have the potential to show things with certainty that they cannot.

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Chapter 1: The importance of the analysis of multiple DNA ancestry tests

The research presented in this thesis is based on the analysis of seven sets of results from various commercialized DNA ancestry tests. The tests are all taken by the author herself, so that the results can be directly compared. As a scholar it is important to look at the way that commercialized DNA ancestry test results are presented to the customer because the presentation of the results, regardless of what the results actually are, can say a lot about what the companies want customers to believe about their product and results. Separate from the results of the tests themselves, this analysis will provide insight into public assumptions surrounding commercialized DNA ancestry tests and what they mean and can say about oneself.

Specifically, it is important to look at these test results in anthropology because of the discipline's background knowledge in multiple relevant fields. The anthropological background in biological science is relevant for this subject because it allows us to have a basic understanding of the scientific significance and potential limits of commercialized DNA ancestry tests. The discipline of anthropology also has a long-standing interest and expertise in how people and cultures define themselves and their relationships to other people and cultures.

Although I am not looking at the science behind the tests themselves in this analysis, understanding fundamentally what they are measuring and observing is important in this situation. Having a background in all of these different areas of expertise allows anthropologists to have a unique perspective, because of the cross-discipline analysis that naturally occurs. In this specific case, I am looking not only at a biological test, but also at how the people that are taking them view these tests. This ability to look across sub-fields within the discipline of Anthropology is what makes Public Issues Anthropology so important and useful.

Currently, the public appears to be using DNA ancestry tests for more than just learning

about their own personal genealogy. As will be shown, customers are also using the tests to contact potential family members, and also to define their ethnicity. My research is important because it explores the boundaries of these tests by comparing and evaluating the various test results. Specifically, I look at the interpretations of the test results presented to the customer from each individual company. I do not look at the actual science behind the tests. This analysis examines what potential tests can show, and hopefully also show the limits of some of these tests if results do or do not overlap. It is important to look at this because if the tests are going to be seen as completely accurate by the public, they should, in theory, be consistent no matter what test is taken by a single person.

The popularity and novelty of commercialized DNA ancestry tests can be seen in the fact that they are appearing in popular culture. On television, TLC has featured a show called “Who Do You Think You Are?” which is in its sixth season and year, and has been renewed for a seventh. This show centers on discovering celebrities’ ancestry, through traditional genealogical means and also through some DNA testing. DNA ancestry testing also recently appeared in the fictional family comedy TV show “Black-ish” on ABC. Additionally, on PBS, there is a TV show, similar to the TLC show, called “Finding Your Roots” that also looks at celebrities’ ancestry. This program focuses more on DNA ancestry rather than having a mainly genealogical focus.

The genetics industry and research into genetics is becoming increasingly popular and is progressing very quickly. The movement of genetics into the commercial and public mainstream is providing a new means for individuals to understand their genetic background, and as anthropologists, we should do our best to understand its implications sooner rather than later, both as academics and as members of the public. The abuse of genetics has created problems in

the past such as eugenics and races, and therefore we should see how these tests and their progress compares, or could cause similar problems. Alternatively, there may be positive outcomes from these tests, which should also be considered, like whether or not they have the potential to refashion or subtilize the term ‘race’.

The possible venue for publication I have chosen for my thesis is *Human Genetics*. I specifically did not want to pick a venue for publication in the area of anthropology, because I wanted to keep with the Public Issues theme and try to involve other disciplines in an anthropological analysis of a traditionally scientific area of interest. Although this would not directly educate the ‘public’ as is regularly defined, it would educate an academic ‘public’ instead. It is my belief that this subject will be of interest to these readers because this research project will hopefully breathe some life into the human/customer aspect of what is otherwise, in the scientific community, known as a strictly scientific endeavor. The idea is that DNA can show ancestry, but questions about why we would need or want this information and whether or not it should be provided to the public do not seem to be broached in this space as often as one might see in an anthropology or other social science publication.

Chapter 2: 4150 Cousins

Part 1: Introduction

The tracing of kin has occurred for centuries within the field of anthropology and has changed throughout the years, as anthropology's idea of what 'kinship' means changes with new technologies, social norms, and beliefs. Commercialized DNA ancestry tests are the newest complication within anthropology's notion of 'kinship', as they challenge traditional notions of the tracing of kin within the discipline of anthropology (Edwards 2012; Parkin & Stone 2004:5-6; Salazar 2012). Additionally, these tests challenge and develop anthropological ideas within the sub-disciplines of archaeology and paleoanthropology, as they claim to be able to map and estimate human migration and how past cultures are related to one another.

Particularly, in the case of DNA ancestry tests, customers of the tests are being encouraged to include new 'relatives' that the testing companies are determining to be possible relatives based on one's genetic codes and Haplogroup. This occurrence will be discussed in more detail below.

There has been other research done in the area of DNA ancestry testing, notably by Paul Brodwin, Carl Elliott, Gísli Pálsson, Jennifer Wagner, and Kenneth Weiss (Brodwin 2002; Pálsson 2012; Wagner 2010; Wagner et al 2002; Wagner & Weiss 2012). Their research will be touched on in more detail below as well.

By looking at multiple tests, I can use the results from each to compare information about what the tests are able to tell a customer. The following presents the results of seven commercialized DNA ancestry tests that are all available to the public, for under \$400 Canadian dollars each. They are not the only tests available, but these tests are chosen because of their popularity, price, and what they claim to be able to report to the customer.

I am able to speak briefly to the role these tests play within anthropological notions of kinship. Specifically, these tests complicate notions that suggest that kinship is either social or biological. The complication comes when the tests report what they have determined to be potential biological kin, with whom the customer shares no social bond. Additionally, I find that none of the tests have the capability to tell a customer decisively where their ancestors lived because of the lack of depth of analysis by which some of the tests determine where a customer ‘matches’, or ‘fits in’ the best. Finally, because of the uncertainties in current genetic science, the ‘results’ provided to the consumer are not at all definitive.

Part 2: Background

Anthropology has long been interested in how people and cultures are related to one another, and how different cultures understand and express those relationships. Where it was once thought that the only ‘relatives’ one had were those who were already known to be directly related to us in a way that could be represented on a genealogical tree or kinship chart, providers of current DNA tests are claiming that they can now discover other relatives who may not fit into traditional genealogical mappings of immediate relations. The tests suggest that certain other customers could be related to the person who has taken the test, because of certain matching genetic markers. However, although these people are all related to the customer in question in some way in the past, there is no way to determine which of these customers are in fact close cousins. Hence, my title, which notes that between the seven tests taken, it is suggested that I could have 4,150 cousins descended from my great-great grandmother, all the way through to my great-great-great-great-great-great grandmother, almost 200 years ago. This problem is explained in more detail below.

These tests are pointing to a new cultural practice in which people are interested in finding out who they are related to genetically and where they come from ancestrally, and also where these relatives fit onto a traditional genealogical chart. This shift in technology/culture is leading to a new way to map human kinship, a direct example of how nature and culture can work together.

The DNA tests that are offered to the public include an analysis of one’s mitochondrial DNA (mtDNA), autosomal DNA, Y-chromosome DNA, or a combination of the above. Men can have all three of these tests performed; however, women can only have their mtDNA and autosomal DNA analyzed because they lack a Y-chromosome. In mtDNA analysis of ancestry,

the company collects the consumer's sample and then determines their genetic code. Specific regions of that code are then analyzed for certain known mutations, or sequences that are different than a sequence which has been determined to be 'normal', or the 'standard sequence' (Budowle et al. 2003:121-122). The placement of these mutations and the letters that are substituted in a person's genetic code that makes them unique are then noted and, once all mutations from an individual are collected, the company can determine one's Haplogroup (Budowle et al. 2003:125). A Haplogroup is a code assigned to a group of persons sharing a certain distinctive set of mutations in their genetic code, in a certain order (Budowle et al. 2003:125). The Haplogroups are sorted into an mtDNA phylogenetic tree, with the origin being a woman dubbed "Mitochondrial Eve" or, as seen in the figure below, "Origin": the earliest set of modern human mtDNA geneticists have determined thus far (see Fig. 1). Each new branch on the figure below indicates a mutation at certain locations of a person's genetic code that was inherited and then passed down to further generations (DNA Ancestry Project 2015). The mutations continue over time, leaving us with the Haplogroups that are seen within the current human population. More of this information will be discussed below regarding my own specific Haplogroup, and what this meant for my results.

In the results, I show that some of the tests also analyze the HVR-1 and HVR-2 locations in my genes. HVR stands for Hypervariable Region, and within one's genome there are two of these regions in one's mtDNA (Family Tree DNA (b)). They actually do not contain any genes, which is why they can change more rapidly, hence the name 'Hypervariable' (Family Tree DNA (b)). Along with the two HVR regions in one's mtDNA, there is also an area called the Coding Region, which contains genes, causing its mutation to occur more slowly (Family Tree DNA (b)).

Once assigned to a Haplogroup, an individual's genetic code is compared to a population that has been constructed by each individual test company to represent a certain population. The comparison of customers' DNA to previous populations in order to determine their ancestry encourages an idea of 'purity'. The companies look for reports, or conduct research to collect genetic information about people who believe their ancestors lived in a certain geographic region for several generations. They then take this information and note the Haplogroups and genetic markers that repeat from these geographic areas. Customers who take the tests are then matched to these various geographic markers, and this is how their geographic ancestry is estimated. The people who represent each geographic area are labeled as being 'purely' from certain geographic regions. This is problematic, as all of the tests use different 'pure' populations as their baseline 'population groups', and this means that when a customer buys a particular DNA ancestry test, the result will differ from what they would have received from other tests. As Marks (2013) discusses, the conclusions from these testing companies come from cultural assumptions about how 'pure' these 'population groups' are. Although the 'population groups' are meant to be biologically constructed, they are in fact subjectively constructed – created by the testing companies – as they are created based on companies' individually constructed databases. The likelihood that one's DNA has the potential to tell the customer that they are ancestrally from Italy may be accurate, but it is known that people from Greece and Tunisia's gene pools overlap with people's genes who are from Italy, blurring the validity of the results (Marks 2013).

Although mtDNA and Y-chromosome DNA can tell the customer about some of their relatives, the tests are limited to looking at only a small portion of a person's lineage. As explained by Carl Elliott and Paul Brodwin (2002:1470):

The problem [with] the mapping [of] Y chromosome and mitochondrial DNA polymorphisms [is that it] will trace only two genetic lines on a family tree in which

branches double with each preceding generation...Continue back...14 generations and the man will be still be connected to only one ancestor in that generation. The test will not connect him to any of the other 16,383 ancestors in that generation to whom he is also related in equal measure.

mtDNA Phylogenetic Tree

The placement of mtDNA Haplogroup I in the mtDNA phylogenetic tree is as follows:

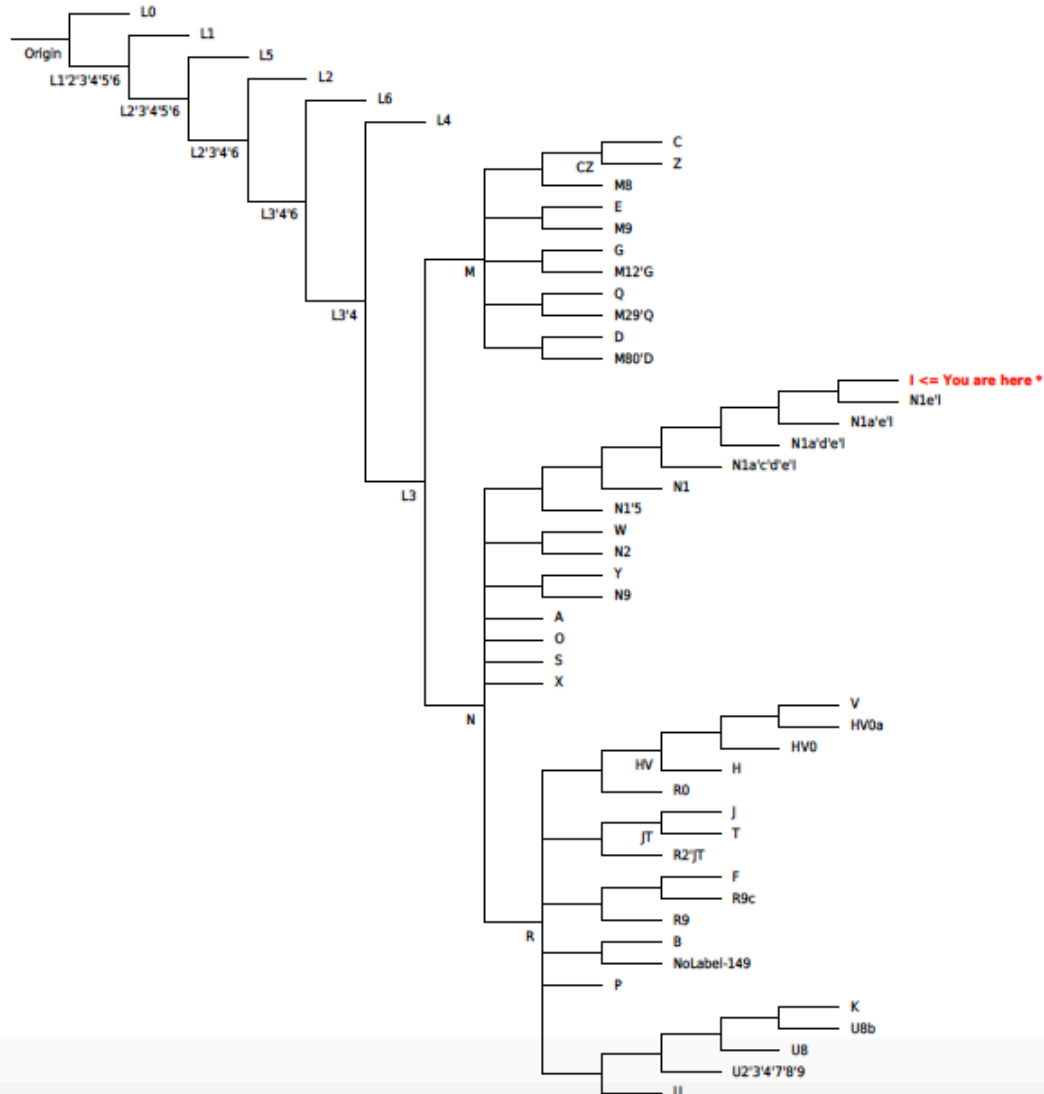


Fig. 1 mtDNA Phylogenetic Tree (DNA Ancestry Project 2015)

Part 3: Method

The most important thing when preparing this research was to ensure that I used a diverse set of DNA ancestry tests from various countries that used different analysis labs, and who reported using different platforms, and offered different price ranges. Since I am a woman, and do not have a Y-chromosome, my only option was to have my mtDNA or autosomal DNA analyzed. Because of financial restrictions, most of the tests I had done only analyzed my mtDNA, as autosomal DNA analysis was significantly more expensive and would have limited the number of tests I could take. In my opinion, this project required a greater number of tests be performed, rather than a greater range of my genetic material be analyzed, because I wanted to compare the results that a consumer receives from different tests, rather than looking at a few tests' scientific approaches in great detail.

The seven tests taken were: DNA Ancestry Project, 23andMe, Oxford Ancestors, ancestryDNA, The Genographic Project, iGenea, and Family Tree DNA. DNA Ancestry Project, a Canadian company, only analyzed my mtDNA. This company provided an emailed report with access to online information, cost \$119 USD (\$143.75 CAD), and its analysis lab was located in British Columbia. 23andMe, an American company who reported their results only through an online platform, a personalized webpage. It only analyzed mtDNA, cost \$199 CAD, and its analysis lab was located in California. Oxford Ancestors, the original commercialized DNA ancestry test, was a British company, and analyzed my mtDNA. It reported information in paper form sent in the mail, cost £199 (\$371.67 CAD), and its analysis lab was located in the United Kingdom. ancestryDNA was a Canadian company, affiliated with the popular genealogy website ancestry.com in the United States. This test provided only web-based results, analyzed mtDNA and autosomal DNA, cost \$149 CAD, and its analysis lab was located in Ireland. The National

Geographic Genographic Project, an American company, tested both mtDNA and autosomal DNA. This test also only reported results online, cost \$199 USD (\$240.39 CAD), and its analysis lab was located in Texas. iGenea, a European company located in Switzerland, had an online report, and also sent an official paper copy in the mail. It analyzed only mtDNA, cost €199 (\$269.32 CAD), and its analysis lab was located in Switzerland. Finally, Family Tree DNA, an American company, analyzed my X-chromosome in order to potentially locate the ‘Warrior Gene’ (Gillet & Tamatea 2012). This test provided the report through email and an online database, costs \$99 USD (\$119.59 CAD), and its analysis lab was located in Texas, at the same facility where The Genographic Project’s DNA was analyzed.

All of the tests were purchased online, and then the sample collection kits were mailed to me from the company. I then either provided a saliva or cheek tissue sample to be analyzed. The results were then sent directly to the laboratory where the samples were analyzed, and their results sent back to the company from which I ordered. Following this, I was either sent the results by email, notified by email that a webpage was ready for me to view, or I was sent the results in the mail.

I confirmed with the Office of Research Ethics that I did not need ethics clearance for this project because I only used my own DNA, thereby giving my own consent by performing the tests. By taking the tests using my own DNA I was trying to see what was and was not included in the results of each of these tests and how the results compared. Each test provided ‘final results’ that were compared and contrasted first and put into groups based on the ‘features’ that were included such as FAQs, personalized results, generic results, health information, maps, certificates, DNA breakdown, scientific information, mundane information, etc. After this initial

analysis, I looked at what was included in the results that pertained specifically to the individual who ordered the test.

Part 4: Results

Of the seven DNA ancestry tests taken, six of the tests were generalized tests, meaning that they identified my Haplogroup and then used that information to determine where my ancestors may have come from geographically. The seventh test, plus one of the generalized tests, searched for a very specific piece of genetic information. The following is a description of the seven tests and the information that each provided to me.

DNA Ancestry Project

The DNA Ancestry Project test assessed the HVR-1 portion of my mtDNA (DNA Ancestry Project 2015). The report arrived via email, in PDF format and included information about the online database that I could access; the specific breakdown of my HVR-1 coding sequence including where my mutations were found; a prediction of my Haplogroup; a description of what is known about my predicted Haplogroup; a phylogenetic tree including my placement on it; a geographic representation of where my Haplogroup resided; a percentage breakdown of the possibility that I was part of specific global populations based on my Haplogroup (e.g. Polish, Romani, Latvian); general information about mtDNA testing and what they look for and why; a FAQs section; and finally a printable, frameable certificate which included my genetic code, and nucleotide mutation (DNA Ancestry Project 2015). The only information that was fully personalized was the page reporting my genetic code, and the certificate at the end of the report. All of the other information would be provided, exactly as it appeared to me, to any other customer who was identified as being part of Haplogroup I (DNA Ancestry Project 2015).

This test, since it only looked at the HVR-1 section of my mtDNA, only analyzed genetic sequences 16001, 16101, 16201, 16301, 16401, and 16501 and the corresponding nucleotide change in those areas (see Fig 2.).

HVR-1 Sequence											
16001	ATTCTAATTT	AAACTATTCT	CTGTTCTTTC	ATGGGGAAGC	AGATTGGGT	ACCACCCAAG	TATTGACTCA	CCCATCAACA	ACCGCTATGT	ATTTCGTACA	
16101	TTACTGCCAG	CCACCATGAA	TATTGTACAG	TACCATAAAT	ACTTGACCAC	CTGTAGTACA	TAAAAACCCA	ATCCACATTA	AAACCCCTC	CCCATGCTTA	
16201	CAAGCAAGTA	CAGCAATCAA	CCTTCAACTA	TCACACATCA	ACTGCAACTC	CAAAGCCACC	CCTCACCCAC	TAGGATACCA	ACAAACCTAC	CCACCCCTAA	
16301	CAGTACATAG	TACATAAAGC	CATTTACCGT	ACATAGCACA	TTACAGTCAA	ATCCCTTCTC	GTCCCATGG	ATGACCCCC	TCAGATAGGG	ATCCCTTGAC	
16401	CACCATCCTC	CGTGAAATCA	ATATCCCGCA	CAAGAGTGCT	ACTCTCCTCG	CTCCGGGCC	ATAACACTTG	GGGTAGCTA	AAGTGAACGT	TATCCGACAT	
16501	CTGGTTCCTA	CTTCAGGGCC	ATAAAGCCTA	AATAGCCAC	ACGTTCCCT	TAAATAAGAC	ATCAGCATG				
HVR-1 Qualified Cambridge Reference Sequence (rCRS) variations											
Nucleotide Position		Region		Variant Type		Nucleotide Change					
16129		HVR-1		Substitution		G>A					
16179		HVR-1		Substitution		C>T					
16223		HVR-1		Substitution		C>T					
16391		HVR-1		Substitution		G>A					
16519		HVR-1		Substitution		T>C					

Fig. 2 HVR-1 Sequence of Danielle Nadeau(DNA Ancestry Project 2015)

Based on these substitutions, this company predicts that I am part of Haplogroup I. This test's description of Haplogroup I is that the woman who founded the group was from the Middle East about 30,000-50,000 years ago, and descended from Haplogroup N. Additionally, most people today in Haplogroup I trace their ancestry back to Europe and the Middle East, but may also be from South Asia and East Africa. Following this, they provided a geographic representation of my entire 'line of descent', including the Origin, followed by Haplogroups L3, N and I (see Fig. 1 for Phylogenetic tree, see Fig. 3 for geographic descent) (DNA Ancestry Project 2015).

The dispersal of Haplogroup I is noted in a chart provided in the report that describes the "Population Distribution Frequency of mtDNA Haplogroup I" in Europe, the Middle East and Central Asia (See Fig. 3) (DNA Ancestry Project 2015). It includes a population, percent of the population that belongs to Haplogroup I, study size, and a reference for this information (DNA Ancestry Project 2015). The greatest percent for Europe was "Polish Roma in Zielona Gora and Nowa Sol, Western Poland" with 7.35% of the population, had a study size of 68 people, meaning that five people in that population were a part of Haplogroup I (DNA Ancestry Project

2015). Here, the report references a report on “Mitochondrial DNA diversity in the Polish Roma”, an article by Malyarchuk BA et al. (2006) from the *Annals of Human Genetics*.

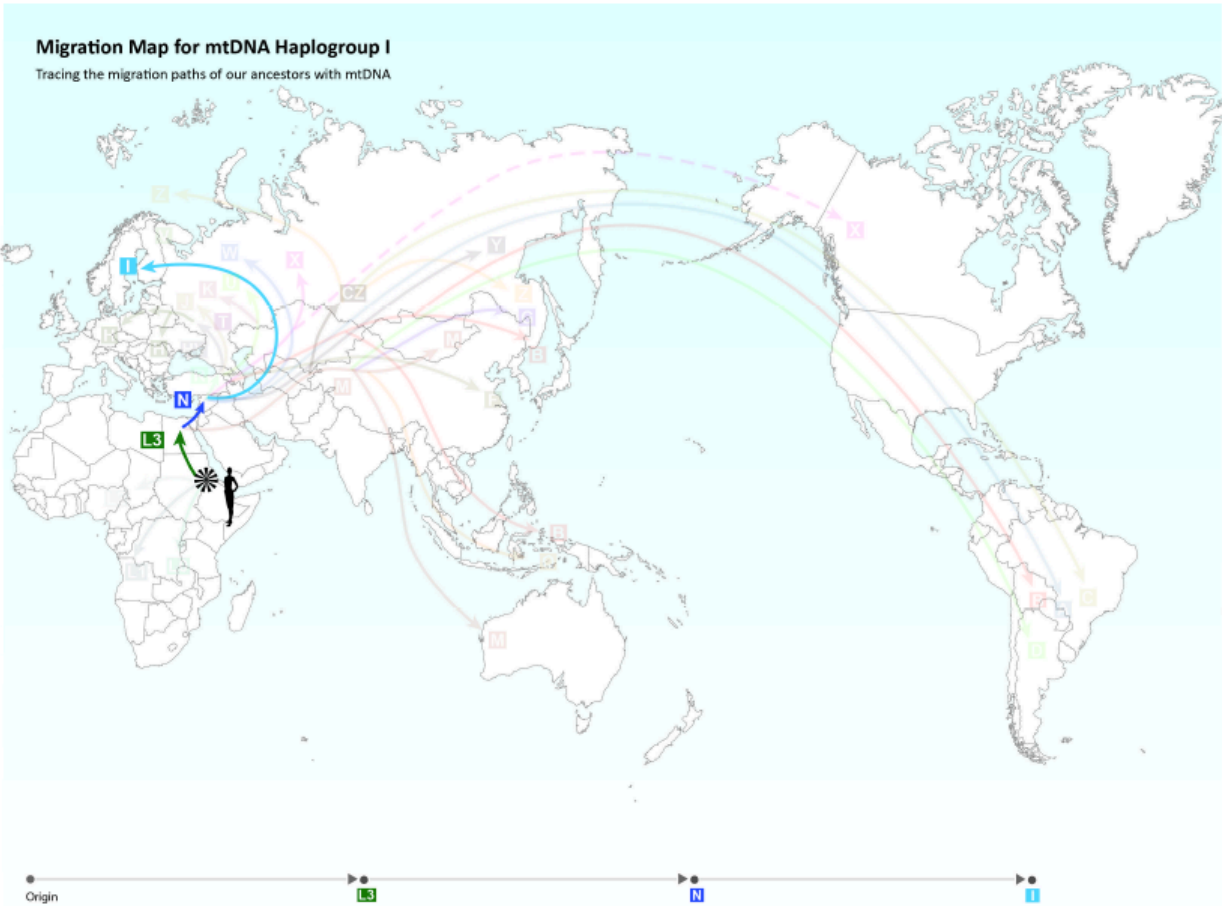


Fig. 3 Migration Map for mtDNA Haplogroup I (DNA Ancestry Project 2015)

In the “Background Information for mtDNA Testing” section of the report, they provided a chart that indicated which Haplogroups, according to their databases and information, belong to which geographic space of the world (see Fig. 4) (DNA Ancestry Project 2015).

Region	Major mtDNA Haplogroups found in region specified
Native Americans	A, B, C, D, X
Oceanic and Aboriginal Australians	O, P, Q, R, S
East Asian	A, B, C, D, E, F, G, M, Y, Z
South Asian (i.e. India)	G, M, W
Europe and Middle East	CZ, H, HV, HV0, I, J, JT, K, R0, T, U, V, W, X
Diverse	N, R
African	L0, L1, L2, L3, L4, L5, L6

Fig. 4 Major mtDNA Haplogroups Found in Specific Regions (DNA Ancestry Project 2015)

The online database for this test provides a personalized webpage, which includes a subscription to their traditional genealogical services. It also provides information about: other members with similar DNA; how DNA has helped archaeologists; indigenous DNA; and Haplogroups. The database the customer has access to containing information about people who have similar DNA shows people's names, with how many DNA marker mutations match your own. One can then send them a message, or view more details about the match (DNA Ancestry Project, 'Genetic Genealogy').

23andMe

I was informed via email that my results were ready to be read, and was guided to a personalized webpage with the following tabs: "Home", "My Results", "Family & Friends", and "Research & Community" (23andMe n.d. (d)). When I clicked on the "My Results" tab, the sub-headings under "Ancestry Overview" were: "Ancestry Composition", "Maternal Line", "Paternal Line", "Neanderthal Ancestry", and "Ancestry Tools" (23andMe n.d.(d)). On the "Ancestry Composition" page I was provided with a map that indicated countries where they believe my ancestors lived (23andMe n.d. (a)). Please see Fig. 5, 6 and 7 for the map views of my ancestry, Fig. 5 is what they considered to be a "Speculative" view of my ancestry, which had a confidence level of 51%; Fig. 6 a "Standard" view, which had a confidence level of 75%; and Fig. 7 a "Conservative" view, which had a confidence level of 90% (23andMe n.d.(a)).

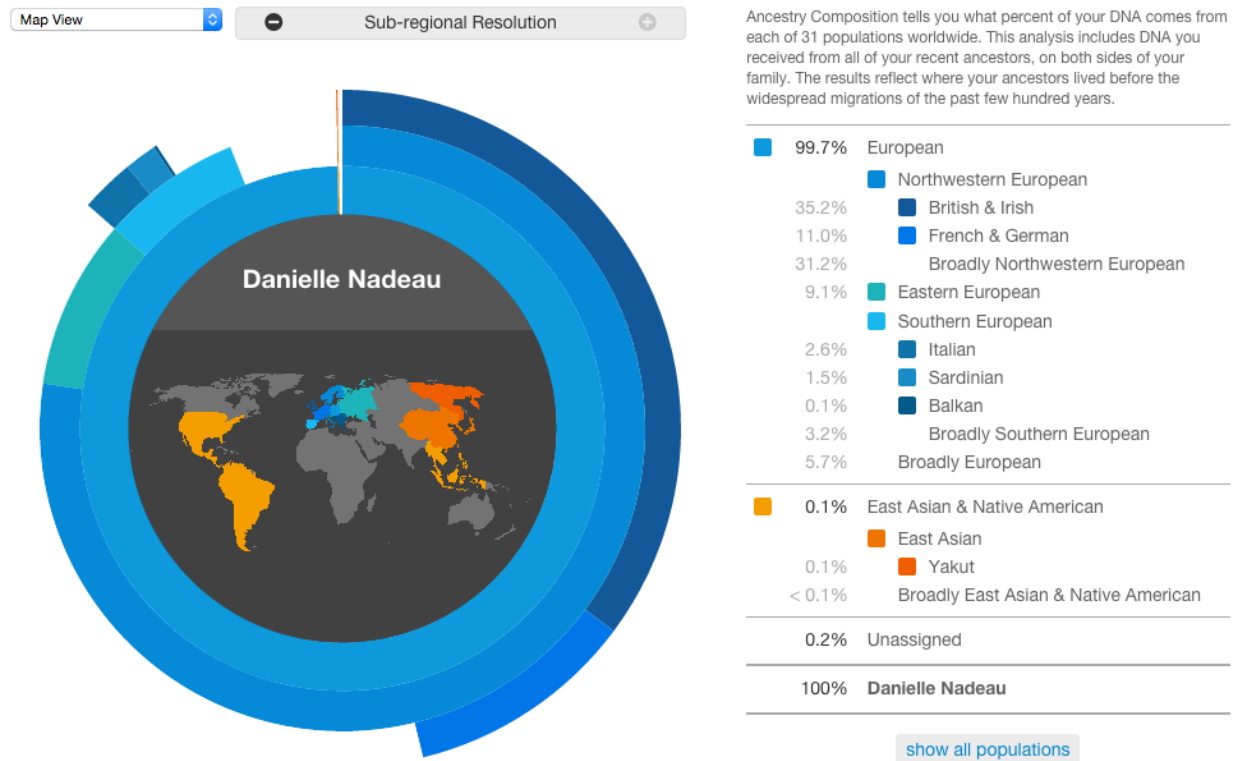


Fig. 5 ‘Speculative’ Estimation of Danielle Nadeau’s Ancestry (23andMe n.d. (a))

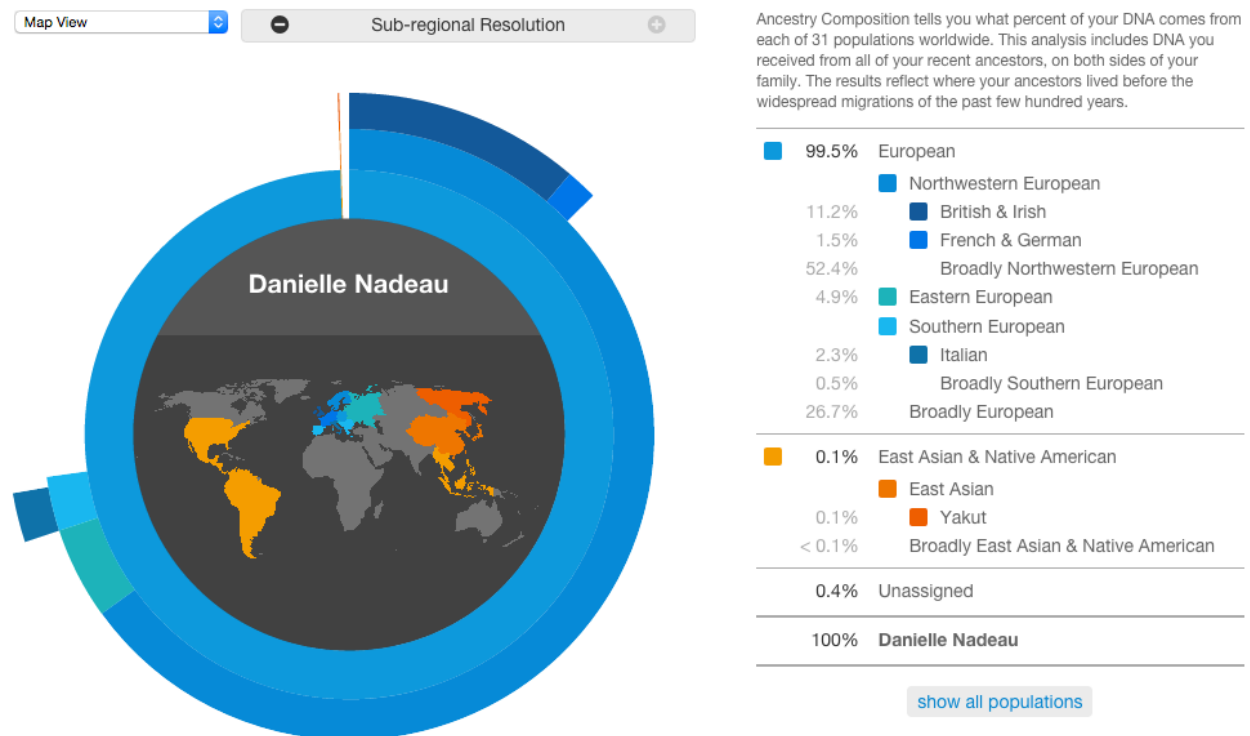


Fig. 6 ‘Standard’ Estimation of Danielle Nadeau’s Ancestry (23andMe n.d. (a))

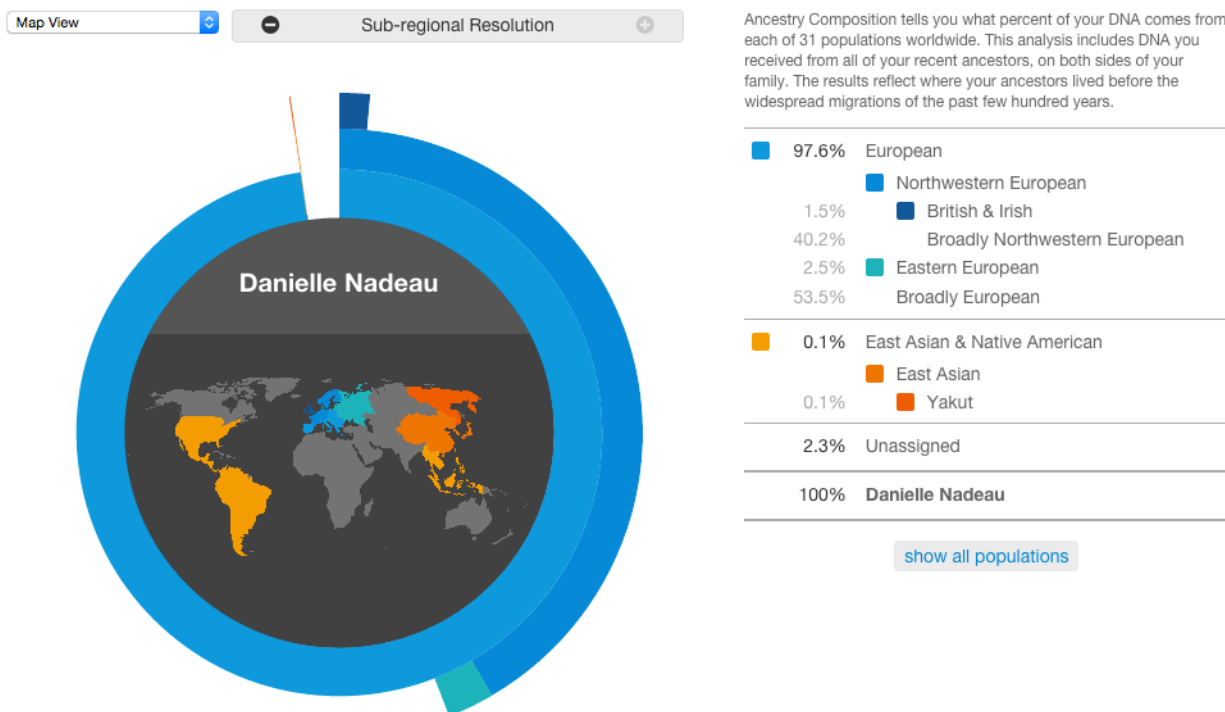


Fig. 7 ‘Conservative’ Estimation of Danielle Nadeau’s Ancestry (23andMe n.d. (a))

Unlike the DNA Ancestry Project test, 23andMe indicated that they base their population percentages on 31 populations but do not cite references anywhere on the website. Also, these geographic results look different than the previous report, because these results indicated the specific geographic country they believe I may be descended from, whereas the geographic representation above only indicated where the founders of each Haplogroup came from (23andMe n.d. (d)).

When I clicked on the sub-heading “Neanderthal Ancestry” I found that they approximate that I am 2.7% Neanderthal. This information was based on Neanderthal DNA (Vi33.16, Vi33.25, Vi33.26) found in the Vindija cave in Croatia. They indicated that the average European user has 2.7% Neanderthal (23andMe n.d. (e)).

Finally, I clicked on “Ancestry Tools” I was brought to a page that offered many activities in which the customer could use their 23andMe results, all designed by other customers

and people from within the company. For example, I could turn my DNA into a melody, or create a Haplogroup Tree Mutation Map (23andMe n.d. (b)).

Under the remaining tabs on the “Home” webpage, there were “Family & Friends” and also “Research & Community”. “Friends & Family” allowed the customer to ‘connect’ with possible relatives that may share a portion of their genetic code. Additionally, the service attempted to predict how far descended this person was from the customer in question (e.g. 4th cousin). I will touch on the importance of this network in more detail below (23andMe n.d.(c)).

Finally, the “Research & Community” tab took the customer to a page that allowed them to answer surveys, and ‘quick questions’ to ‘help’ 23andMe with their research and predictions (23andMe n.d. (f)). Additionally, the customer can find out about discoveries made because of other customers’ answers to the previously mentioned surveys and questions, current research projects, and forums of questions and answers contributed by the consumers who have used the page (23andMe n.d. (f)).

There was no FAQ section, or explanation of how the DNA sample was analyzed (23andMe n.d. (f)). There was a breakdown of one’s genetic code that one could download, but it downloaded into an Excel spreadsheet which was completely unintelligible to someone outside the company and who was not a geneticist (see Fig. 8 for a small example of what was included in the document).

6163	kgp10409159	4	T	T
6164	kgp10412223	7	G	G
6165	kgp10412739	14	C	C
6166	kgp10414554	4	-	-
6167	kgp10428523	2	-	-
6168	kgp10431183	13	T	C
6169	kgp10445883	16	T	T
6170	kgp10455993	4	T	T
6171	kgp10460254	17	C	C
6172	kgp10473467	10	T	C
6173	kgp10475253	18	G	G
6174	kgp10479159	12	C	C
6175	kgp10495478	5	-	-
6176	kgp10500578	15	A	A
6177	kgp10519840	13	A	G
6178	kgp10525713	18	A	A
6179	kgp10532243	10	T	T
6180	kgp10546200	21	T	G
6181	kgp10546654	2	A	A
6182	kgp10557595	3	A	G

Fig. 8 DNA Analysis of Danielle Nadeau (23andMe n.d. (g))

Oxford Ancestors

This company is the original commercialized DNA ancestry testing company. The founder, Bryan Sykes, wrote the book The Seven Daughters of Eve (2002), which traces all living humans back to seven women, “Clan Mothers”, that he named for the Haplogroup they founded: Ursula, Xenia, Helena, Velda, Tara, Katrine, Jasmine, and Ulrike. My result for this test indicates that I am from Xenia’s clan, or Haplogroup X. Sykes hypothesizes that there are 29 other “Clan Mothers” alive at some point in history – including Haplogroup I – but, that they do not have any living descendants bearing their genetic mutation (Oxford Ancestors 2015).

The results of this test were sent by mail, and were received in hard copy in an official folder including: an official chart of “The World Clans” with my name and clan mother’s name, and my genetic mutation locations printed at the bottom; an official chart of where I was located within the “Seven Daughters of Eve”, with the same information as previously noted printed at the bottom of the page; a map showing the migration patterns of each of the Seven Daughters of Eve, showing that Xenia occupied most of Europe and part of the Middle East, lining up with information that had been provided to me by the other companies regarding Haplogroup I, and

where they believe my ancestors came from; a pamphlet explaining the process of breaking down one's genetic code and how the results were interpreted, and a pamphlet about each of the Seven Daughters of Eve with a 'story about their lives' that Sykes wrote about in his book The Seven Daughters of Eve (Oxford Ancestors 2015; Sykes 2002).

In the pamphlets that came with the results, there were also instructions about how to log onto their online database to find relatives from the same "Clan Mother". This service was the same as the ones mentioned above, where one is pre-matched with people who share some sort of genetic code that indicates you come from a certain "Clan Mother" (Oxford Ancestors 2015).

ancestryDNA

This test is affiliated with the popular genealogical website ancestry.ca. This test notified me via email that my results were ready to be seen, and provided a link to a personalized webpage (ancestryDNA n.d.). However, the DNA feature on this website was extremely new and the customized page was very simple, suggesting that the DNA test was just an accessory to their main product, the traditional genealogical mapping (ancestryDNA n.d.). On the "Home" page of the webpage, there was an "Ethnicity Estimate" (ancestryDNA n.d.). This is the only test I took that used the term "ethnicity" instead of "geography" (ancestryDNA n.d.). When one clicks on the "Ethnicity Estimate" one was brought to a new webpage that showed a map, and highlighted one's "Ethnic Ancestry", which was the same as the geographic distributions provided by the other tests (see Fig. 9) (ancestryDNA n.d.).

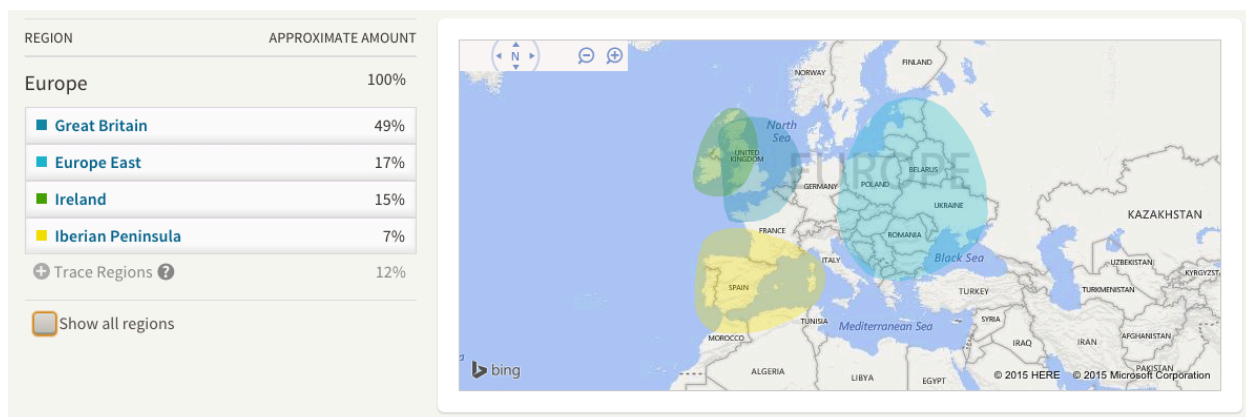


Fig. 9 Danielle Nadeau’s Ethnicity Estimate (ancestryDNA n.d.)

This service did not provide Haplogroup information, and did not show the customer anything about their genetic code. There was some information about how one’s “Ethnicity Estimate” was determined: it was noted that they compare the consumer’s DNA sample to over 4,000 other DNA samples from twenty-six global regions, mostly collected by the Sorenson Molecular Genealogy Foundation (Ball et al. 2013). This website also provided the service to meet potential relatives and offered the ability to add them to the genealogical tree that you have constructed on the company’s sister website (ancestryDNA n.d.).

Genographic Project

This test is run by National Geographic and is presented to the customer as a way to help people better understand the human journey rather than their personal ancestry, unlike the others. I was not notified by any means when my results were ready to be viewed; instead one must log in to the site, usually multiple times after the sample is sent away to see if the results are available. The information was provided only using a personalized web page through National Geographic’s website (The Genographic Project n.d. (b)).

Once one has logged in, the first information available to the consumer was a section asking if the consumer would like to know more information about their ‘ancestral journey’.

They provide a very comprehensive guide to how a consumer's DNA is analyzed, what the test is able to tell the customer, which relatives can be seen with the specific tests, and how they turn the information from genetics into geography. National Geographic uses 43 reference populations to determine a consumer's ancestry (The Genographic Project n.d. (a)). They do not provide information about how big the reference populations are, or where they got their data.

The next information available to the customer was "Hominin Ancestry". My personal results are 3.5% Neanderthal and 2.8% Denisovan. The percent of Neanderthal that this test reported is greater than the 23andMe results, which only reported 2.7%. Also, the "Average" numbers that each test reports should be in the average consumer is also different. 23andMe reported a 2.7% average, and The Genographic Project reports only 2.1% (The Genographic Project n.d. (d)). The Genographic Project does not provide any explanation of where they get the information about Neanderthal or Denisovan ancestry to compare to modern humans.

Following "Hominin Ancestry" I was provided with my "Deep Ancestry" information, which is from between 1,000-100,000 years ago. "Deep Ancestry" denotes my Haplogroup, or the point at which my ancestors' genes mutated on my maternal side and have not yet changed. They categorize me as Haplogroup I4A, a sub-sub-category of Haplogroup I. The Genographic Project provided additional information about this Haplogroup, stating that they have determined that Haplogroup I developed about 20,850 years ago in West Africa, and the population moved into, and spread throughout Europe (The Genographic Project n.d. (c)).

Next was my "Regional Ancestry", which dates from 5,000-10,000 years ago. The Genographic Project reports that I am 43% Northern European, which could be from the UK, Denmark, Finland, Russia, or Germany; 38% Mediterranean, which could be from Sardinia, Italy, Greece, Lebanon, Egypt, or Tunisia; and finally 18% from Southwest Asia, which could be

from India, neighboring regions to India, Tajikistan, or Iran, and could also be from Eastern Europe or North Africa. My breakdown was then compared to the previously mentioned reference populations. For instance, they showed that my results were closest to that of their example British population (see Fig. 10).

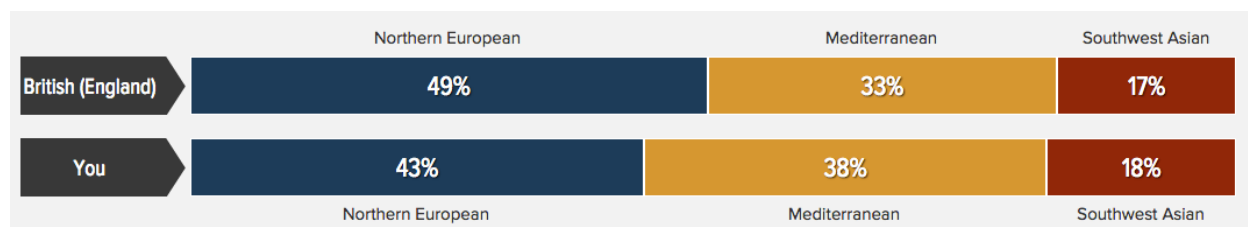


Fig. 10 Danielle's DNA compared to a 'Typical British Person' (The Genographic Project n.d. (e))

They also indicated that my "Regional Ancestry" was comparable to their reference German population (see Fig. 11) (The Genographic Project n.d. (e)). It was not explained why they have determined that I was more closely related to someone from England than Germany, although, as you can see, it looks as though I match better with the DNA from Germany.

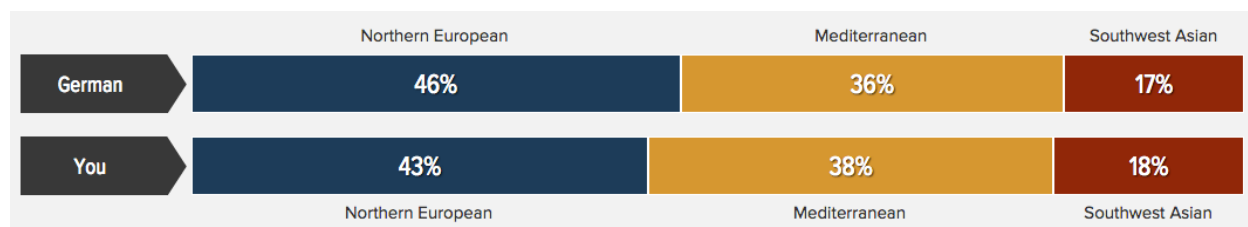


Fig. 11 Danielle's DNA compared to a 'Typical German Person' (The Genographic Project n.d. (e))

This was the only information provided to the consumer from this test that was personal, although, there was a massive database of information regarding how they determined one's genetic code, what they were doing with the information for research purposes, etc. Also, unlike all of the other tests thus far, there was no network to find potential relatives. Also, there was no information whatsoever regarding my genetic code, or mutation locations anywhere on the web page.

iGenea

This test looked at my autosomal DNA, and also looked to see if I had ‘Viking genes’. I was mailed a hard copy of my results, and they also created a personalized webpage for my results. In the hard copy of my results I was provided with an official certificate of my results including my HVR-1 and HVR-2 mutation locations and letter changes, that they used to determine that I am part of Haplogroup I, from the ancient tribe of the Celts, not the Vikings, and that my region of origin is North-West Europe and Ireland. This company describes Haplogroup I as being my pre-historical ancestry, being a Celt as my ancestry from antiquity, and being from North-West Europe and Ireland as my ancestry from the Middle Ages. The hard copy also included information about DNA ancestry analysis, answering questions such as “What is DNA?”, “How can DNA tell anything about my Origins?”, etc. The online database information was the same information that was found on the hard-copy certificate. There was also no database to connect with other members through this test (iGenea 2015).

Family Tree DNA

This test determines if I have any ‘Warrior Genes’. Briefly, the “Warrior Gene” is a specific Monoamine oxidase (MOA) gene, specifically, MAO-A30bp-rpt, which corresponds with higher risk-taking, and more aggressive behavior (Lea & Chambers 2007). MOAs are “...enzymes responsible for breaking down the neurotransmitters —serotonin, dopamine, and adrenalin—and are therefore capable of affecting mood...” (Lea & Chambers 2007). The MOA gene is located in the X chromosome so men have one but women have two. If one has the “Warrior Gene”, the test run by Family Tree DNA will show a value of 3; ‘normal’ variants of the MOA gene will be represented as 3.5, 4, 4.5, or 5 (Family Tree DNA n.d. (b)). Therefore, women will have two resulting numerical representations, either two ‘normal’ variant numbers, two 3 results, or one 3 result and one ‘normal’ variant. The result of my test was that I had two 3

results so both of my X-chromosomes contain the “Warrior Gene” variant (Family Tree DNA n.d. (b)).

Test	DNA Ancestry Project	23andMe	Oxford Ancestors	ancestryDNA	Genographic Project	iGenea	Family Tree DNA
Type of DNA Tested	mtDNA HVR-1	Not listed	mtDNA HVR-1 and HVR-2	mtDNA and autosomal DNA	mtDNA and autosomal DNA	mtDNA HVR-1 and HVR-2	X-Chromosome
Price of the Test (in CAD on date of purchase)	\$143.75	\$199	\$371.67	\$149	\$240.39	\$269.32	\$119.59
Nationality of the Company	North American	American	British	Canadian	American	Swiss	American
Type (Generalized/ Specialized)	Generalized	Generalized	Generalized	Generalized	Generalized	Generalized and Specialized (Viking genes)	Specialized (Warrior Gene)
Results Delivery Method	E-mailed PDF, online profile page	Personalized webpage	Hard copy	Personalized webpage	Personalized webpage	Hard copy and personalized webpage	Personalized webpage
Haplogroup Assigned	I	None	Clan of Xenia	None	I4a	I	N/a
Genetic Code Included	Yes, full code with mutations noted	Yes, but incomprehensible	No	No	No	No	No
Information About Testing Available	Yes, included in PDF and online	Yes, limited online	Yes, limited in hard copy results bundle	Yes, limited online	Yes, plethora online	Yes, limited in hard copy results bundle	Yes, limited to compressed blurb about Warrior Gene
Relatives' Finder Service	No	Yes	Yes	Yes	No	No	N/a
Geographical Representation Included	Yes, of Haplogroup, not personal	Yes, personal, regional, by country	Yes, of clan mother, not personal	Yes, personalized	Yes, personalized	Yes, personalized	N/a
Notes				Used the term "ethnicity"			

Table 1 Comparison of all DNA ancestry tests

As one can see in Table 1, of the companies that report Haplogroups, all of the companies except Oxford Ancestors, who reported that I am a member of Haplogroup X, reported that I am a member of Haplogroup I. As mentioned, Oxford Ancestors presupposes that Haplogroup I existed, but that no one survives from that descent line (23andMe n.d. (a); The Genographic Project (c); iGenea 2015; Oxford Ancestors 2015). Oxford Ancestors does not explain why all the other companies apparently disagree with their interpretation.

The geographic information included with each of the tests did not show exactly the same geographic ancestry. DNA Ancestry Project (2015), and Oxford Ancestors (2015) both included geographic information, but both show geographic information about Haplogroup distribution, and were not personalized to the consumer. The areas that these tests said the Haplogroup distribution moved to, were both Northern and Eastern Europe, and Oxford Ancestors (2015) also noted that the distribution of my assigned Haplogroup was distributed into Eastern Europe, the Middle East and Russia, and potentially to North American indigenous populations.

Of the personalized geographic information provided by the remaining tests – 23andMe (n.d. (a)), ancestryDNA (n.d.), and The Genographic Project (n.d. (c), (d)) – the geographic information they provided regarding my ancestry are all similar but not exactly the same. 23andMe (n.d. (a)) reported, conservatively, that I am: 97.6% European, 0.1% East Asian & Native American, and 2.3% unassigned. ancestryDNA (n.d.) reported that I am: 100% European, with many percentages coming from various European ranges. They did not report any ancestry from East Asian or Native American, like 23andMe did. Finally, The Genographic Project (n.d. (d)) reported that I am 43% Northern European, 38% Mediterranean, and 18% from Southwest Asia, which matched the 23andMe (n.d. (a)) results better than the ancestryDNA results, but are still very different.

Part 5: Discussion and Implications

This research did not involve interviewing customers using DNA tests, but from the test companies' advertisements it seems possible to infer what the companies believe their customers would like to see in the results. The content of the ads seems to suggest that the customers are looking for easy-to-understand information about who their ancestors and relatives are, and where they came from.

In anthropological terms, and according to what can be inferred from the companies' advertisements, the customer is seeking kin. Based on the information provided in the reports, the companies also seem to believe that the customer does not care about their genetic code or mutations. This is based on the fact that of the seven tests taken, only two [DNA Ancestry Project (2015) and 23andMe (n.d. (c)))] included my genetic code in the results, and 23andMe's method of delivery of the genetic code was extremely hard to track down and was completely incomprehensible to a non-expert (see Table 1). However, none of the companies, in my opinion, do an adequate job of explaining what a person's genetic code means, how Haplogroups are determined, how statistically certain the laboratory is of one's genetic code determination, or the likelihood that a person could have a genetic anomaly or mutation that would alter the results from what is normally expected. Although they all provided information about the test that was conducted to determine one's DNA mutations, and some provided information about one's Haplogroup, statistical probabilities of the tests were not addressed by any of the seven companies (see Table 1).

Additionally, the matching of potential relatives to customers implies a kin relationship between those people but, as an anthropologist, this is troubling to me. The test companies seem to infer that customers want to know about immediate kin, so they encourage the customer to

believe that these people are more closely related than can be known to be with any certainty. For example, in the 23andMe (n.d. (c)) results, they list people with “Shared Segments” to my profile and go so far as to label them with potential genealogical labels (see Fig. 12).









	Danielle Nadeau Female	You	I4	UPDATE YOUR PROFILE
	Male	3rd to 4th Cousin 0.98% shared, 2 segments	Canada windsor cousineau trahan K2a R1b1b2a1a2f*	Public Match Send a Message
	Female	3rd to 4th Cousin 0.71% shared, 3 segments	K1b2b	Send an Introduction
	Female	3rd to 4th Cousin 0.61% shared, 3 segments	United States Eastern Europe PILKINGTON ON MY MOTHERS... McIntosh H5a1	Public Match Send a Message
	Female	M 3rd to 4th Cousin 0.54% shared, 3 segments	K1a1b2a	Send an Introduction
	Female	3rd to 4th Cousin 0.54% shared, 3 segments	H4a1	Send an Introduction
	Female	3rd to 5th Cousin 0.54% shared, 3 segments	H	Send an Introduction
	Female	3rd to 5th Cousin 0.54% shared, 3 segments	United States *Hull Canada, Levi Canada, Detr... Bastien Bernier H2a1 Emond	Send an Introduction

Fig. 12 Example of some ‘Potential Cousin’ matches of Danielle Nadeau (23andMe n.d. (c))

All of these people, and more, are listed on my profile as ‘potential’ 3rd to 5th cousins. A 3rd cousin would be descended from a common great-great grandparent who, assuming generations are approximately 25.5 years, would have been born approximately 125 years ago; a “4th cousin” descended from a common great-great-great grandparent born approximately 150 years ago, and a “5th cousin” descended from a common great-great-great-great grandparent born approximately 175 years ago.

So, as a customer I have a problem: do I add all these people to my genealogy chart (Devine n.d.)? And as an anthropologist I have a similar problem: are these people actually my kin? If one believes traditional theories of evolution, then every human is related to every other,

as we have all evolved from the same ancestor if you go far enough in the past. So, yes, technically these people are all related to me in one way or another. But, how far in the past are we related? Can 'our' notion of cousinhood be met just because of a genetic relationship, or is something more needed?

The answer is that we do not know, and we cannot determine any of the answers to those questions with any certainty at this point because current genetic literature has not definitively answered these and many other pertinent questions in the genetics field. As one can see, the person with whom I am most closely matched above shares only 0.98% of my explored genetic code (23andMe n.d. (c)). In the human body there are an estimated 19,000-20,000 protein-coding genes, one's mtDNA accounts for 37 of these genes, one's mtDNA has 16,600 base pairs, and only portions of these are looked at when determining one's ancestry (Ezkurdia et al. 2014). 23andMe looks at all three of these regions of one's mtDNA, but also looks at autosomal DNA, and analyzes all of this information using a microchip which can recognize single nucleotide-polymorphisms (SNP), otherwise known as nucleotide mutations (International Society of Genetic Genealogy Wiki n.d.). With this chip, 23andMe is able to look at 10% of the total SNP's in the body on all of our genes, and they are also able to see approximately 15% of our mtDNA (illumina n.d.). Thus, the people included in Fig. 12 as cousins actually have less than 1% of a match to me, and that is only determined based on the limited amount of genetic code that is examined.

As mentioned above, all of these tests use various databases of the population's genetics to reach these conclusions. Therefore, all of the tests use different sets of data. All three tests have their own databases that they have built individually, using different participants and methods. Additionally, they build their databases based on information from these projects, and

rely upon people who claim to know their genealogy well enough to definitively know their ancestry. They then analyze these people's genes and see what genetic mutations or Haplogroups are duplicated, and use this as the foundation for their individual databases.

Commercialized DNA ancestry tests are very intriguing to customers because of their appeal to the 'unknown'. The companies advertise, and suggest, that they can tell one about their 'unknown' family history. A few anthropologists who have looked at commercialized DNA ancestry tests in their work have discovered that the public expects more from these tests than is provided. Gísli Pálsson (2012) did a study similar to this one looking at just two tests, deCODEme and 23andMe. He argued that these tests blur the lines between experts and lay people, allowing the lay to see information previously only available to experts. It is precisely this notion that I am arguing is to the deficit of the customer. The companies are not providing enough information about exactly what their tests can show the customer. They do explain how the laboratories analyze the sample, in order to create the results the report to the customer, but they do not tell the customer about how precise their results are based on current genetics research and the scope of the tests they are performing.

Wagner and Weiss (2012) also looked at attitudes to DNA ancestry tests. They found that the public who had not participated in DNA ancestry tests thought that the people who chose to take the tests had some genetics knowledge to begin with, and also that these tests could prove someone's race in a legal situation. Additionally, they found that people who had experience taking the tests liked to communicate with others in their identified Haplogroup, but mainly wanted the information for genealogical purposes. This indicates a disconnect between customers who have or have not taken the tests, how companies are targeting customers, and

what their thoughts are regarding what customers want to get out of a test (Wagner & Weiss 2012).

DNA ancestry testing has been considered in the field of anthropology, mainly concerning race and ethnicity; however, few prior studies have considered what the tests mean for the analysis of ancestry or kinship. Also, there seems to be an apparent disconnect between the customer and company in more than one aspect of the test. As others have shown, customers who have and have not taken the tests all seem to have different ideas of what the tests are useful for, and how they should be used (Pálsson 2012; Wagner & Weiss 2012). Also, as I have shown, these tests cannot, with absolute certainty, tell the customer what their ancestry is, as their individual analyses are subjectively constructed by each individual testing company, which is why different results are determined based on each kind of test one takes.

In my opinion, the most troublesome aspect of the results are from the 23andMe, Oxford Ancestors and ancestryDNA databases used to ‘find relatives’, who are other customers that have the same Haplogroup, or another genetic identifier (23andMe n.d. (c); ancestryDNA n.d.; Oxford Ancestors 2015). The test results imply that the customer is closely related to the people it matches them with, or that they share some sort of history.

With the current information available in the field of genetics, these matches are not, by any means, absolute truths. This is because people can share genetic markers, but may not necessarily be within the same Haplogroup. Additionally, there are potential anomalies that could occur within someone’s genetic code, which are essentially unidentifiable to a testing company who is only looking at certain portions of one’s genetic code makeup. For example, paternal mtDNA inheritance can occur, where a person can have inherited maternal and paternal mitochondrial DNA from their parents (Schwartz & Vissing 2002). From their perspective, they

are asked to determine a person's genetic code and then identify nucleotide variations; their job is not to determine if a customer has any genetic anomalies that could alter the results.

Based on this research, I believe that it is important for the public to clearly understand that these test results cannot, with absolute certainty, tell us who we are and are not related to in any sort of genealogical way, nor can they definitively tell us from what ancestral population(s) we are descended. Most of the tests do not provide the customer with their own genetic code, and although some of the tests provide geographic information to the customer, it is unclear how definitive those results are. Additionally, the public needs to understand that genetic science is new, and ongoing. Therefore, commercials or advertisements implying that a customer could definitively change their identity based on the test's results or even meet 'relatives' are essentially making a false claim.

The public needs to know this so an educated decision can be made when determining whether or not to take these tests. There is also a lot of room for future research in this subject area, since these tests intersect with so many anthropological subjects including kinship, race, ethnicity, biology, and archaeology. Additionally, research into identity formation of the customer based on the results could also be explored. I believe that as our knowledge of genetics changes, so too will the conclusions that have been drawn regarding DNA ancestry tests, as their purpose, and certainty will inevitably change over time as technology advances.

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